



juvenile primary lateral sclerosis

Juvenile primary lateral sclerosis is a rare disorder characterized by progressive weakness and tightness (spasticity) of muscles in the arms, legs, and face. The features of this disorder are caused by damage to motor neurons, which are specialized nerve cells in the brain and spinal cord that control muscle movement.

Symptoms of juvenile primary lateral sclerosis begin in early childhood and progress slowly over many years. Early symptoms include clumsiness, muscle weakness and spasticity in the legs, and difficulty with balance. As symptoms progress, the spasticity spreads to the arms and hands and individuals develop slurred speech, drooling, difficulty swallowing, and an inability to walk.

Frequency

Juvenile primary lateral sclerosis is a rare disorder, with few reported cases.

Genetic Changes

Mutations in the *ALS2* gene cause most cases of juvenile primary lateral sclerosis. This gene provides instructions for making a protein called alsin. Alsine is abundant in motor neurons, but its function is not fully understood. Mutations in the *ALS2* gene alter the instructions for producing alsin. As a result, alsin is unstable and is quickly broken down, or it cannot function properly. It is unclear how the loss of functional alsin protein damages motor neurons and causes juvenile primary lateral sclerosis.

Inheritance Pattern

When caused by mutations in the *ALS2* gene, juvenile primary lateral sclerosis is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- JPLS
- juvenile PLS
- PLSJ
- primary lateral sclerosis, juvenile

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Juvenile primary lateral sclerosis
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1853396/>

Other Diagnosis and Management Resources

- GeneReview: ALS2-Related Disorders
<https://www.ncbi.nlm.nih.gov/books/NBK1243>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Health Topic: Degenerative Nerve Diseases
<https://medlineplus.gov/degenerativenervediseases.html>
- Health Topic: Paralysis
<https://medlineplus.gov/paralysis.html>

Genetic and Rare Diseases Information Center

- Juvenile primary lateral sclerosis
<https://rarediseases.info.nih.gov/diseases/4485/juvenile-primary-lateral-sclerosis>

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke
<https://www.ninds.nih.gov/Disorders/All-Disorders/Motor-neuron-diseases-Information-Page>
- National Institute of Neurological Disorders and Stroke: Spasticity Information Page
<https://www.ninds.nih.gov/Disorders/All-Disorders/Spasticity-Information-Page>

Educational Resources

- Disease InfoSearch: Primary lateral sclerosis juvenile
<http://www.diseaseinfosearch.org/Primary+lateral+sclerosis+juvenile/5952>
- MalaCards: primary lateral sclerosis, juvenile
http://www.malacards.org/card/primary_lateral_sclerosis_juvenile
- Merck Manual Consumer Version: Amyotrophic Lateral Sclerosis and Other Motor Neuron Diseases
<http://www.merckmanuals.com/home/brain-spinal-cord-and-nerve-disorders/peripheral-nerve-disorders/amyotrophic-lateral-sclerosis-and-other-motor-neuron-diseases>

Patient Support and Advocacy Resources

- Motor Neurone Disease Association (UK)
<http://www.mndassociation.org/>
- Muscular Dystrophy Association
<https://www.mda.org/>
- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/primary-lateral-sclerosis/>

GeneReviews

- ALS2-Related Disorders
<https://www.ncbi.nlm.nih.gov/books/NBK1243>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22juvenile+primary+lateral+sclerosis%22+OR+%22primary+lateral+sclerosis%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28juvenile+primary+lateral+sclerosis%5BALL%5D%29+OR+%28jpls%5BTIAB%5D%29+OR+%28plsj%5BTIAB%5D%29%29+NOT+%28IAHSP%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- PRIMARY LATERAL SCLEROSIS, JUVENILE
<http://omim.org/entry/606353>

Sources for This Summary

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<https://www.ncbi.nlm.nih.gov/books/NBK1243>
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Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/condition/juvenile-primary-lateral-sclerosis>

Reviewed: July 2013

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services